



**PARENT INFORMATION for presumptive  
positive newborn screening results for  
MCAD  
(Medium Chain Acyl CoA Dehydrogenase  
Deficiency)**

When your baby was born a small sample of blood was taken from your baby's heel to test for certain diseases. These diseases usually can not be found simply by looking at your baby, so that is why the blood test is so important.

The results of your baby's screening test were "presumptive positive" for a disorder called MCAD. It is important to remember this is only a screening test. A new specimen will be collected from your baby and a new test will be done to help diagnose your baby, or to rule out this disorder.

**? What does a "positive" screen mean?**

A small percent of all babies screened will be identified as "positive" on screening, but are later found to not have the disorder. This is just a screening test and further testing must be done to determine if your child has this disorder, or to rule it out as a "false positive". Keep in mind that the new test could show that your baby doesn't have MCAD.

**☛ What is the next step?**

It is important to follow your baby's doctor's instructions for getting a new specimen for further testing as soon as you receive them. In the meantime, before the results of the additional test(s) are available, your doctor will talk with you about your baby's current health. The doctor may advise you in steps that you may take to reduce the risks of problems occurring for your baby if he/she does have MCAD. Since MCAD is a rare metabolic disease, your doctor may consult with or refer you to a pediatric metabolic specialist.

**? What exactly is MCAD?**

MCAD is a disorder of the body's fatty acid metabolism. People with MCAD lack the enzyme necessary to metabolize some fats into sugars. Problems can occur during periods of fasting, such as when too much time occurs between breast or bottle feeding, or during periods of illness when vomiting or diarrhea occur. In these cases the baby's blood sugars can drop to dangerous levels. This metabolic crisis can lead to seizures, coma, and even death. If the baby survives the metabolic crisis, they may have severe developmental disabilities.

Finding out early if your baby has MCAD means you and your baby's doctor can take steps to prevent this!



**What can I do to prevent these effects?**

The most important thing you can do is to be sure your baby does not have any periods of **fasting (going without any feedings) for more than 4 hours at a time**. Usually newborns will need to be fed more frequently than this, so you should not worry as long as your baby doesn't go for more than 4 hours at a time without a feeding. Secondly, report to your baby's doctor immediately, if your baby should show any signs of illness such as fever, vomiting, or diarrhea. Third, your baby's doctor may prescribe a metabolic supplement called "carnitine". If she/he does, it will be important to follow the doctor's instructions.

You have probably already done this, but now is also a good time to be sure your newborn is covered by your health insurance provider. Most health insurance covers all or part of the medical monitoring needed for MCAD.

For more information talk with your baby's doctor, or you can contact the Nebraska Newborn Screening Program at (402) 471-0374 for information on Pediatric Metabolic Specialists available in Nebraska.